Congenital Adrenal Hyperplasia (CAH)

Congenital adrenal hyperplasia is a family of inherited autosomal recessive disorders of adrenal steroidogenesis. It results from a defect in any of the five enzymes needed to synthesize cortisol from cholesterol in the adrenal gland, but about 80% of all cases are due to deficiency of the enzyme 21-hydroxylase (21-OH). About 15% of cases are due to 11β-hydroxylase deficiencies. The goal of newborn screening for CAH is to rapidly identify affected infants in order to prevent death from adrenal crisis, shock or its sequelae, and incorrect sex assignment in female newborns.

Estimated Incidence (MI):	1:20,000
Laboratory Screening Test:	Fluoroimmunoassay is used to detect elevated 17- hydroxyprogesterone (OHP)
Timing of Test:	Interpretation of values is based on birth weight and age at the time of specimen collection. False positive may occur if sample is collected before 24 hours of age.
Feeding Effect:	None
Transfusion Effect:	None
Steroid Effect:	Chronic use of dexamethasone in the mother during pregnancy can falsely depress 17-OHP levels, which can cause a false negative result in an affected newborn.
Confirmation:	All strong and borderline positive tests are referred to the Endocrine Follow-up Program (EFUP) (734) 647- 8938. The EFUP coordinates follow-up for infants with suspected CAH. Diagnosis and treatment is provided through a network of pediatric endocrinologists throughout the state.
Treatment:	Glucocorticoid is used to replace deficient cortisol while suppressing ACTH overproduction. Salt-retaining hormones may also be used. Early intervention and surgical correction of ambiguous genitalia allow normal puberty, fertility and childbearing in females. Children with CAH require careful regulation of hormone treatment during illnesses and growth periods.

For further information, contact the Newborn Screening Program
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